NLRP7 gene

NLR family pyrin domain containing 7

Normal Function

The *NLRP7* gene provides instructions for making a protein that is thought to play a role in turning off (inactivating) genes based on which parent the copy of the gene came from, a phenomenon known as genomic imprinting. For most genes, both copies of the gene (one copy inherited from each parent) are active in all cells. For a small subset of genes, however, only one of the two copies is active; for some of these genes, the copy from the father is normally active, while for others, the copy from the mother is normally active. The NLRP7 protein is thought to be involved in imprinting multiple maternal genes in egg cells (oocytes) that contribute to a developing embryo, ensuring that the genes will be inactive; the corresponding paternal genes are active.

The NLRP7 protein is likely involved in cell growth and division (proliferation) and cell maturation (differentiation). Research suggests that the NLRP7 protein also plays a role in inflammation and other immune responses by regulating the release of an immune protein called interleukin-1 beta.

Health Conditions Related to Genetic Changes

recurrent hydatidiform mole

More than 50 mutations in the *NLRP7* gene have been found to cause recurrent hydatidiform mole. A hydatidiform mole occurs early in pregnancy when an embryo does not fully develop and the placenta develops abnormally. The placenta, a solid structure in the uterus that normally provides nutrients to a growing fetus, is dysfunctional and appears as numerous small sacs, often described as resembling a bunch of grapes. There is a risk of the hydatidiform mole becoming cancerous. Women who have repeated instances of hydatidiform mole have a condition called recurrent hydatidiform mole. *NLRP7* gene mutations are responsible for recurrent hydatidiform mole in 75 percent of women with this condition.

The *NLRP7* gene mutations that cause recurrent hydatidiform mole lead to production of a protein with impaired function or prevent production of any protein at all. As a result, it is likely that the normal imprinting process is impaired, leading to the activation of many maternal genes that should not be expressed. The overexpression of multiple genes during embryonic development results in poor development of fetal and placental tissues characteristic of a hydatidiform mole. Women with recurrent hydatidiform mole have *NLRP7* gene mutations in all of the body's cells, including oocytes, so a hydatidiform mole will develop in each pregnancy that occurs with

those egg cells. Additionally, *NLRP7* gene mutations result in slowed release of interleukin-1 beta, which disrupts the body's normal immune response that would recognize a hydatidiform mole as a non-growing pregnancy or foreign tissue and signal the body to remove it. Instead, the hydatidiform mole remains in the body.

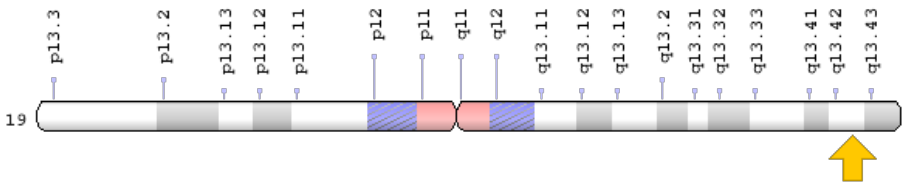
other disorders

Some variations (polymorphisms) in the *NLRP7* gene are associated with an increased risk for early pregnancy loss in females who have the genetic change in one copy of the *NLRP7* gene in each cell. The pregnancy losses include multiple miscarriages and nonrecurrent (sporadic) hydatidiform moles. Unlike women with recurrent hydatidiform mole (described above), women with these polymorphisms are able to have normal pregnancies. *NLRP7* gene variants do not appear to be a major factor in pregnancy losses in the general population. Genetic and environmental factors all play a part in the complexities of embryonic development.

Chromosomal Location

Cytogenetic Location: 19q13.42, which is the long (q) arm of chromosome 19 at position 13.42

Molecular Location: base pairs 54,923,509 to 54,965,184 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CLR19.4
- NACHT, leucine rich repeat and PYD containing 7
- NACHT, LRR and PYD containing protein 7
- NACHT, LRR and PYD domains-containing protein 7
- NALP7
- NLR family, pyrin domain containing 7
- NOD12

- nucleotide-binding oligomerization domain protein 12
- nucleotide-binding oligomerization domain, leucine rich repeat and pyrin domain containing 7
- PAN7
- PYPAF3
- PYRIN-containing Apaf1-like protein 3

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28NLRP7%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- NLR FAMILY, PYRIN DOMAIN-CONTAINING 7
  http://omim.org/entry/609661

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_NLRP7.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=NLRP7%5Bgene%5D
- HGNC Gene Family: NLR family
  http://www.genenames.org/cgi-bin/genefamilies/set/666
- HGNC Gene Family: Pyrin domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/994
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=22947
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/199713
- The Registry of Hereditary Auto-Inflammatory Disorders Mutations: NLRP7
  http://fmf.igh.cnrs.fr/ISSAID/infevers/search.php?n=8
- UniProt
  http://www.uniprot.org/uniprot/Q8WX94

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